ABSTRACT

Title: Methods

This invention relates to polymorphisms and novel sequence in the human pyruvate dehydrogenase E1α (PDH E1α) gene. The invention also relates to methods and materials for analysing allelic variation in the PDH E1α gene, and to the use of PDH E1α polymorphism in the diagnosis and treatment of diseases in which modulation of pyruvate dehydrogenase activity could be of therapeutic benefit, such as diabetes, asthma, obesity, sepsis and peripheral vascular disease. In particular, the invention is based on the discovery of a nucleotide polymorphism in the 3' untranslated region (3'UTR) of the human PDH E1α gene. In addition, we disclose the sequence of intron 7 of the human PDH E1α gene and identify two polymorphisms within intron 7.